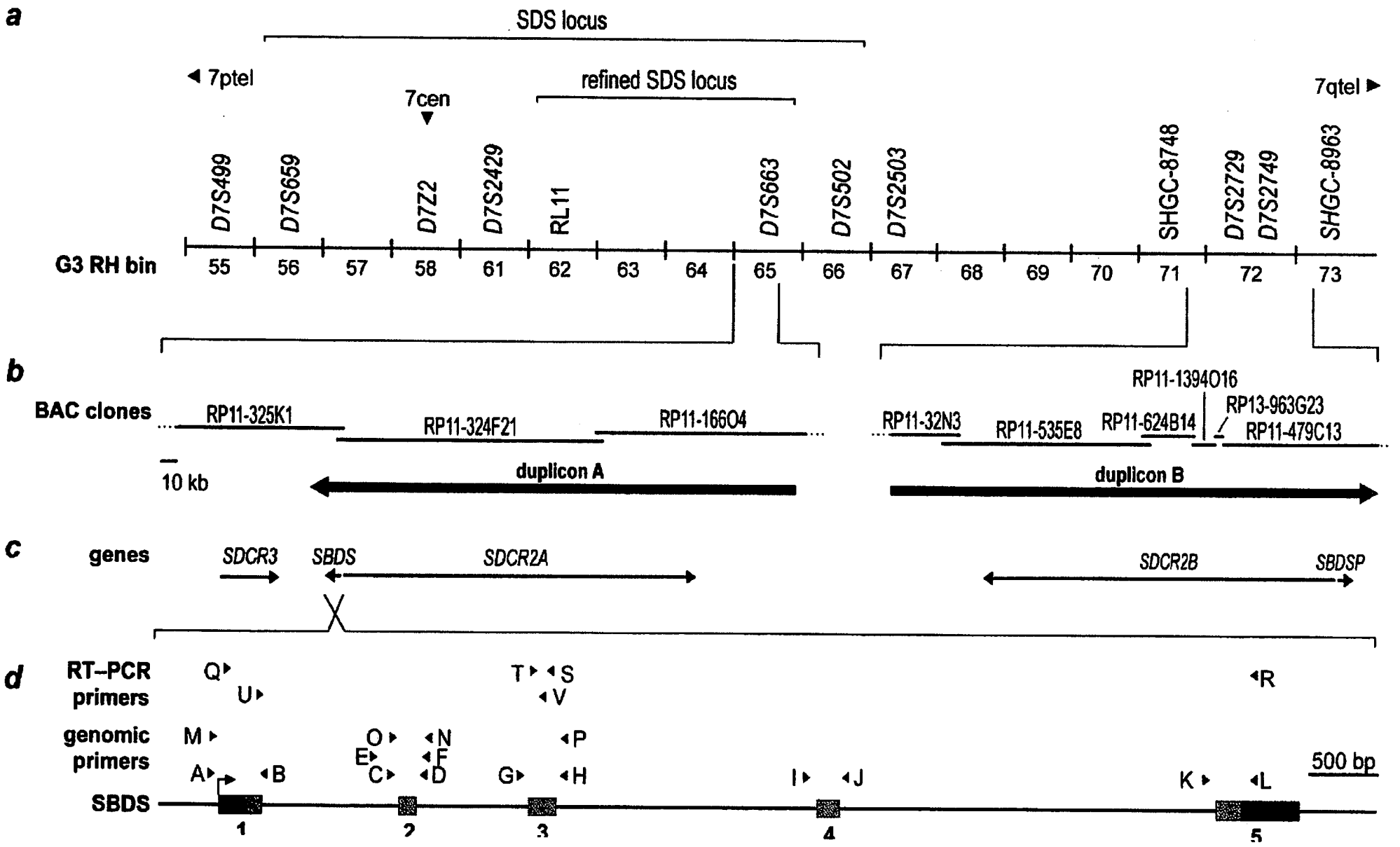
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7cen
refined SDS locus
7qtel
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D7S659
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RL11
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D7S502
D7S2503
SHGC-8748
D7S2729
D7S2749
SHGC-8963
G3 RH bin
55 56 57 58 61 62 63 64 65 66 67 68 69 70 71 72 73
b
BAC clones
RP11-325K1
RP11-324F21
RP11-166O4
RP11-1394O16
RP11-32N3
RP11-535E8
RP11-624B14
RP13-963G23
RP11-479C13
10 kb
duplicon A
duplicon B
c
genes
SDCR3
SBDS
SDCR2A
SDCR2B
SBDSP
d
RT–PCR primers
genomic primers
Q
U
T
S
V
R
M
O
N
P
E
F
A
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D
G
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I
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K
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SBDS
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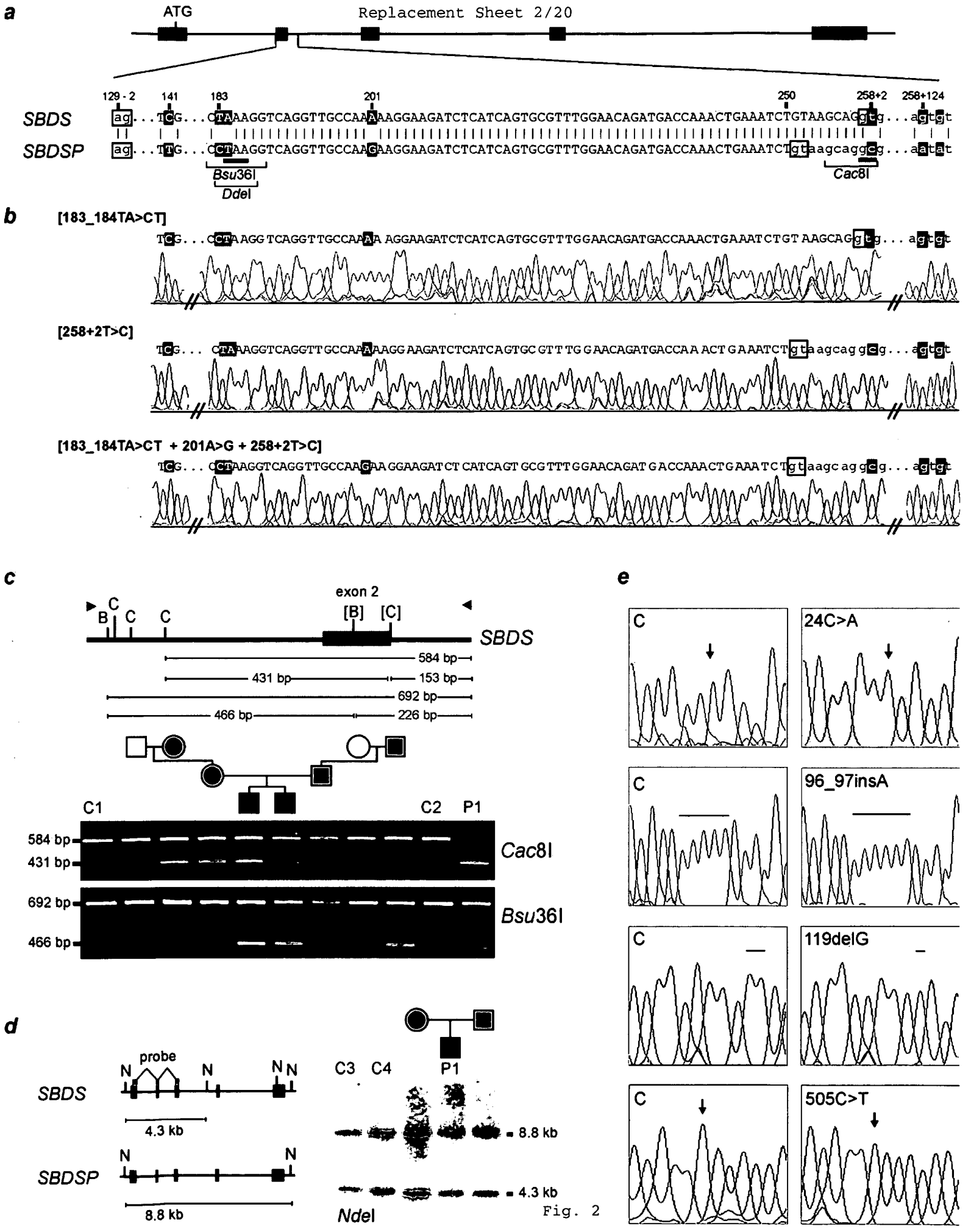
2
3
4
5

Fig. 1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b
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c
exon 2
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431 bp
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692 bp
466 bp
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C1
C2
P1
Cac8I
Bsu36I
d
probe
SBDS
SBDSP
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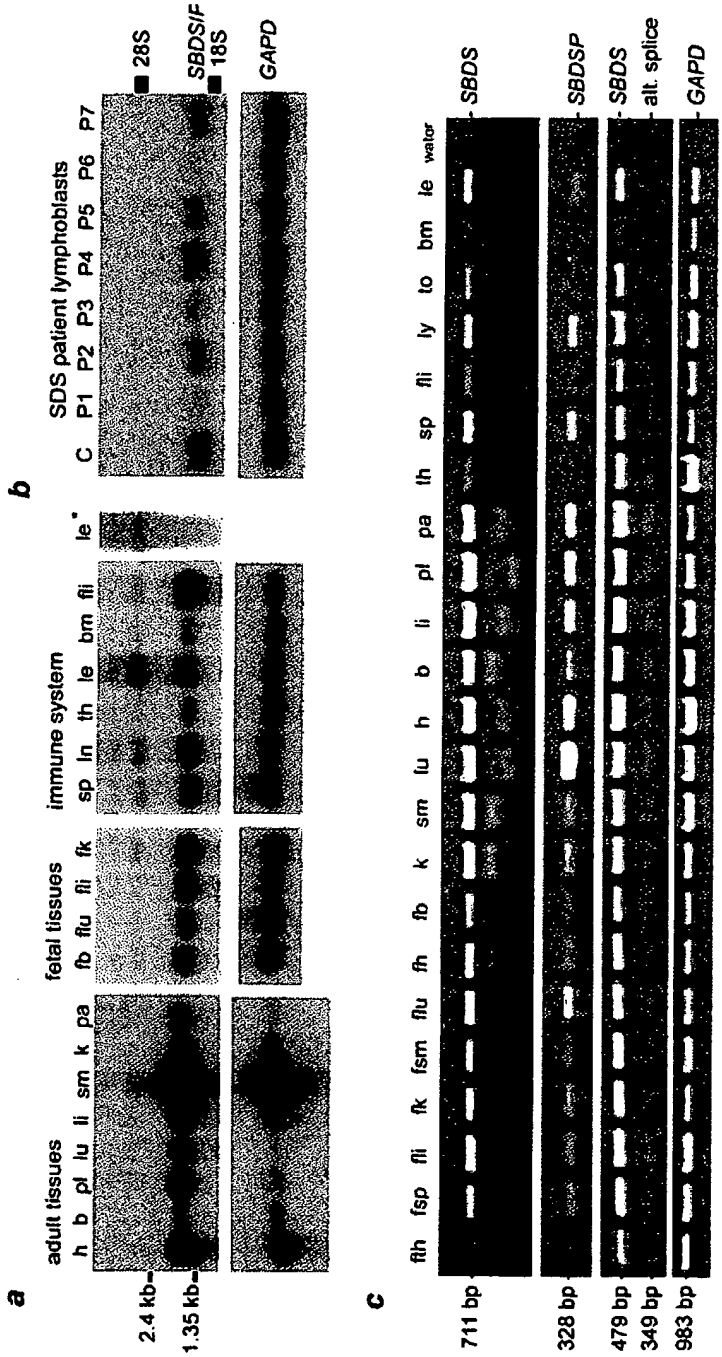
Fig. 2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C P1 P2 P3 P4 P5 P6 P7
28S
SBDS/F
18S
GAPD
c
flh fsp fli fk fsm flu fh fb k sm lu h b li pl pa th sp fli ly to bm le water
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alt. splice
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Fig. 3

SEQ ID NO: 2

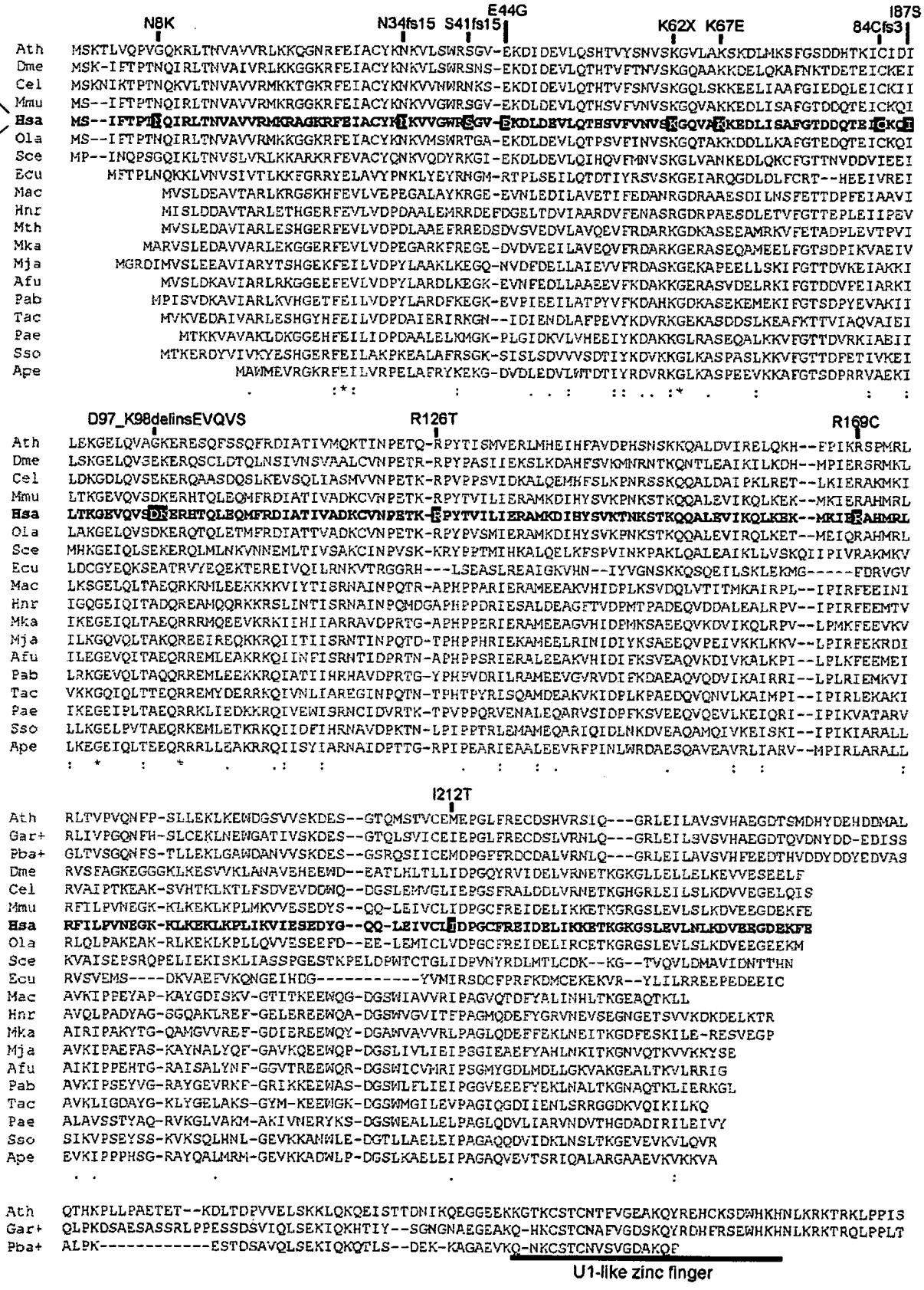


Fig. 4

SBDS Exon 1: (SEQ ID NO: 35)

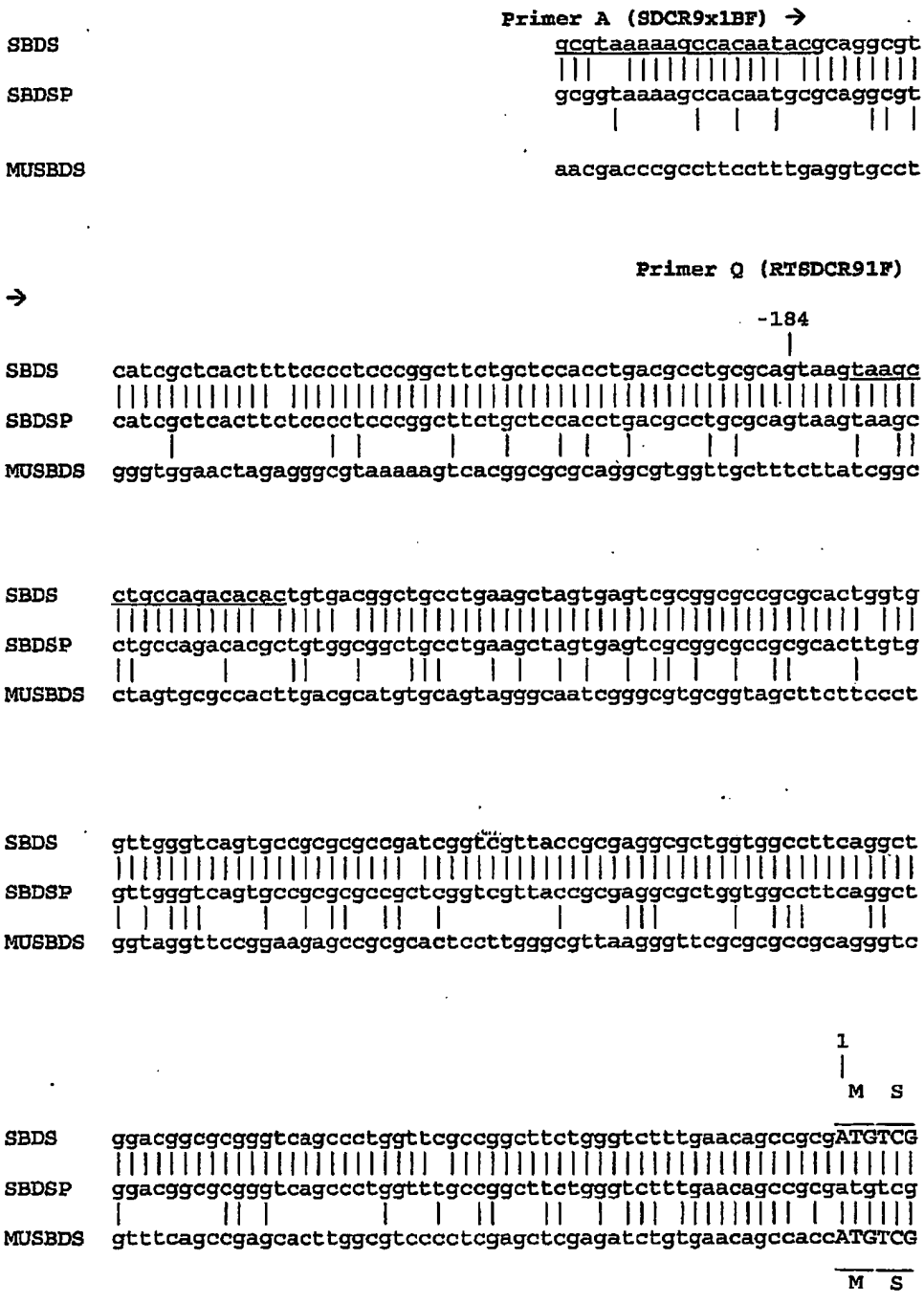
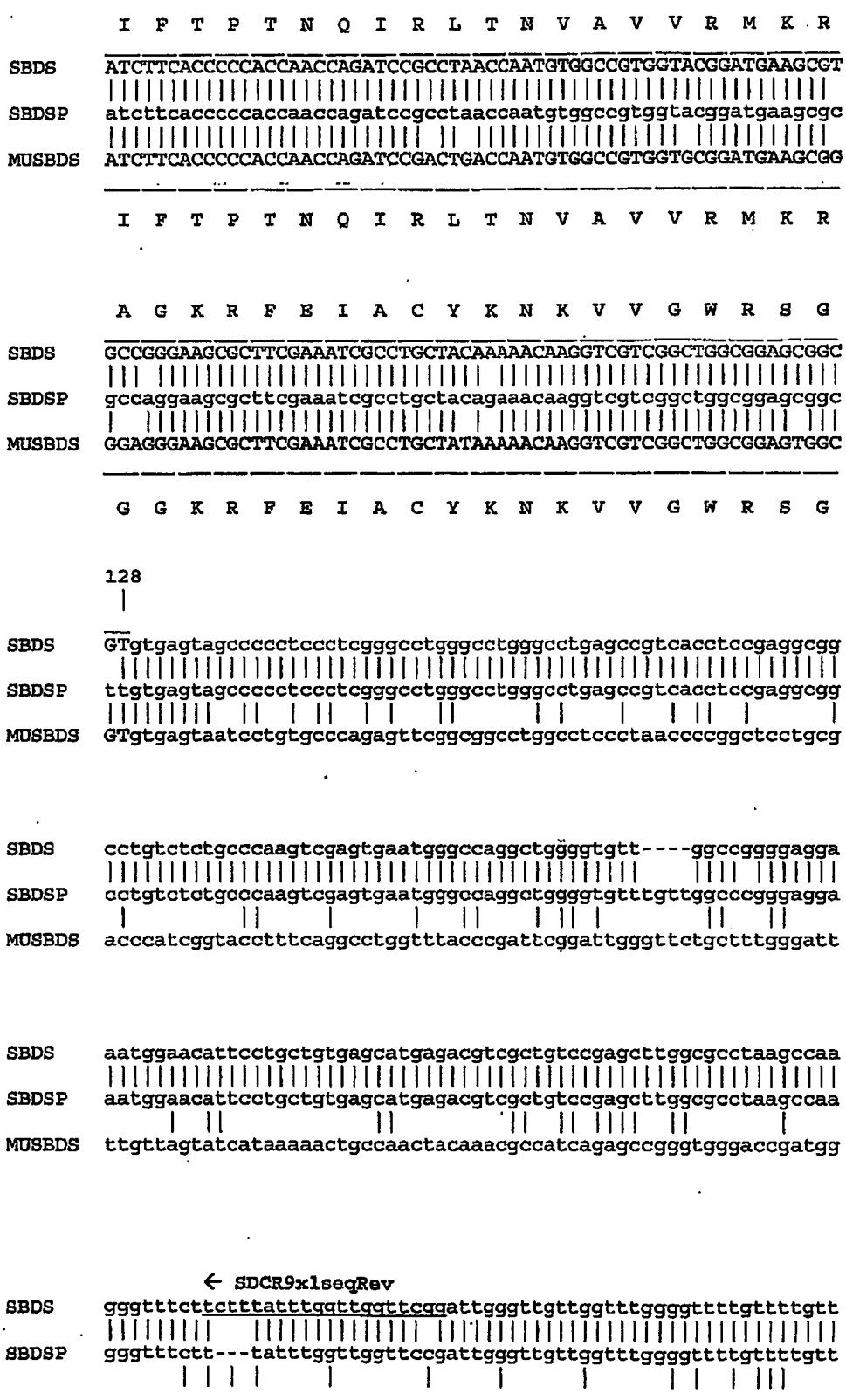


Fig. 6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I F T P T N Q I R L T N V A V V R M K R
A G K R F E I A C Y K N K V V G W R S G
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G G K R F E I A C Y K N K V V G W R S G
128
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SBDSP ttgtgagtagcccccctccctcgggcctgggcctgggcctgagccgtcacctccgaggcgg
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SBDSP aatggaacattcctgctgtgagcatgagacgtcgctgtccgagcttggcgcctaagccaa
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← SDCR9x1seqRev
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Fig. 6 (cont.)

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        ||||||||||||||||||||||||||||||| |||||||||||||||||||||||||||
SBDSP   ggtgtcataaaagctgcagccaagaaatctcataattgtggtccttttcctagaataatg
        |    | |  ||  ||       |       | ||  ||||     |        ||
MUSBDS  gccagcctggaacgtgtgtgtgtgtgtatgtgtatgtgtgtgttgtgtgtgtatgtgt

                    ← Primer B (SDCR9x1BR)
SBDS    atggctgagaacctagtcttacgaatactgtcatag
        ||||||||||||||||| || ||||||||||||||
SBDSP   atggctgagaacctagtgttccgaatactgtcatag
        |||  || |             ||        |
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SBDS Exon 2: (SEQ ID NO: 36)

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        |||||||| ||||||||| ||||||||||||||||||||||||||||||||||||||||
SBDSP   aaatggtagggcaaatacagttctgagttttgaaaatgttccctcaggccgatgcgggca
          |  ||       |     |||  |    || | |              |  |
MUSBDS  gtagtgtcttcgctactgccatctagggacagatattccaggacagaagaaacaccactc


SBDS    gttcacttgaggccaggagttcgaggccagcctggccaacatgaaaccccatctctacta
        | ||||||||||||||||||||||||||||||||||||||||||||||| ||||||||||||
SBDSP   gatcacttgaggccaggagttcgaggccagcctggccaacatgaaacaccatctctacta
           |       ||       |    |  |       |      |||          |
MUSBDS  cccaccacaccctgagtttccttacataaaacaatgatgtagtttttccctctgtggtga


SBDS    aaaatacaaagttagccgggtgtggtggcgcatgcctgtaatcccagttactcaggaggc
        |||||||||| ||||||||||||||||||||||||||||||||||||| ||||||||||||
SBDSP   aaaatacaaaattagccgggtgtggtggcgcatgcctgtaatcccagctactcaggaggc
        |       ||   ||          | ||   |  |    |              |   |
MUSBDS  agtgggagaatccagatactgtccttcgcaggtagccaccagagagagagtgtggtgtgt


SBDS    tgaggcgggagaatcacttgaacccgggaggctgaggttacagtgacccgagatcgcgcc
        |||||| |||||||||||||||||||||||| || ||| |||||| ||||||||||||
SBDSP   tgaggcaggagaatcacttgaacccgggaggcggacgttgcagtgagccgagatcgcgcc
           | | |||| |    |               |  ||||   ||         |
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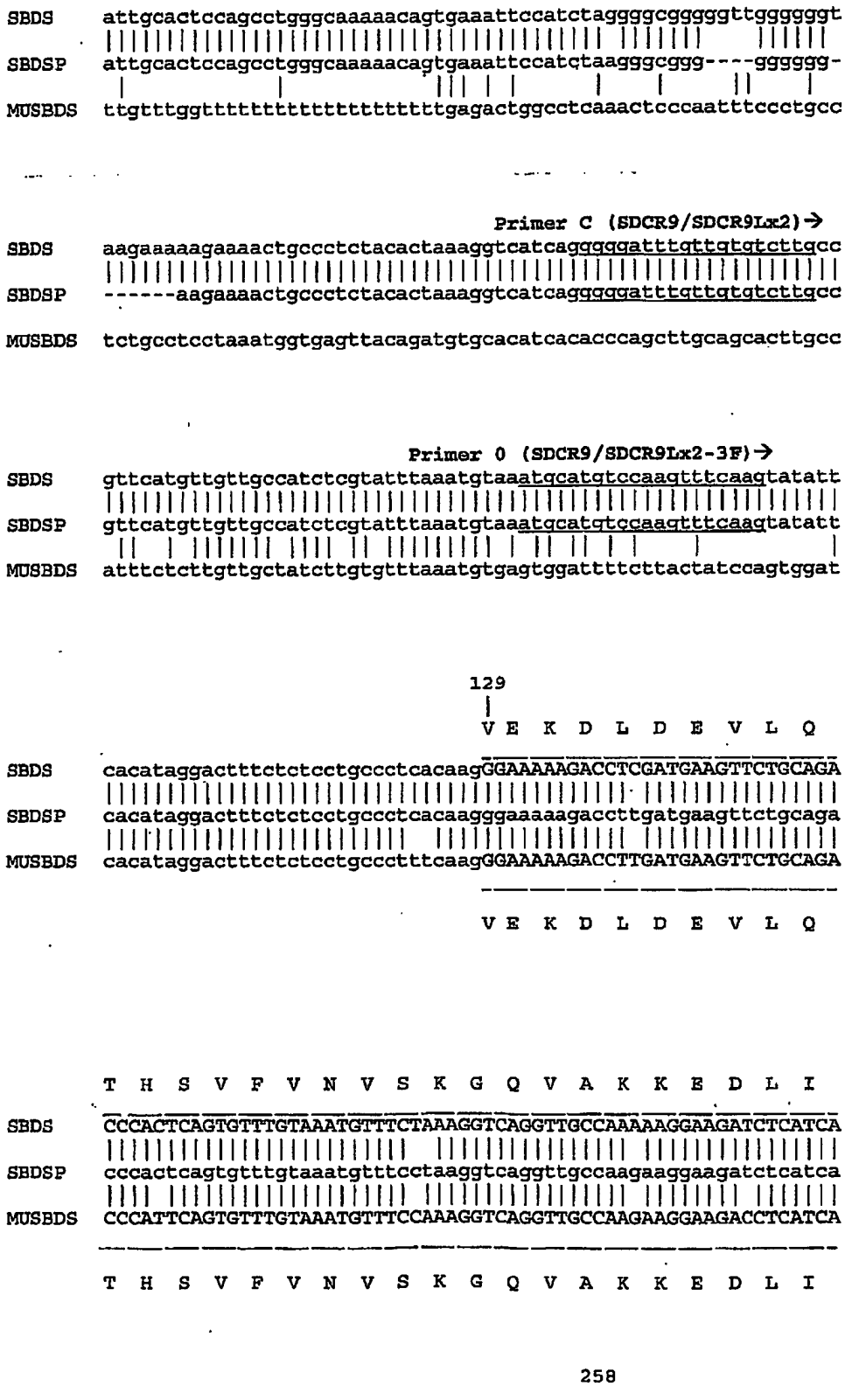
Fig. 6 (cont.)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MUSBDS ttgtttggtttttttttttttttttttttgagactggcctcaaactcccaatttccctgcc
Primer C (SDCR9/SDCR9Lx2)→
SBDS aagaaaaagaaaactgccctctacactaaaggtcatcaggggggatttgttgtgtcttgcc
SBDSP ------aagaaaactgccctctacactaaaggtcatcaggggggatttgttgtgtcttgcc
MUSBDS tctgcctcctaaatggtgagttacagatgtgcacatcacacccagcttgcagcacttgcc
Primer 0 (SDCR9/SDCR9Lx2-3F)→
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129
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V E K D L D E V L Q
T H S V F V N V S K G Q V A K K E D L I
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SBDSP cccactcagtgtttgtaaatgtttcctaaggtcaggttgccaagaaggaagatctcatca
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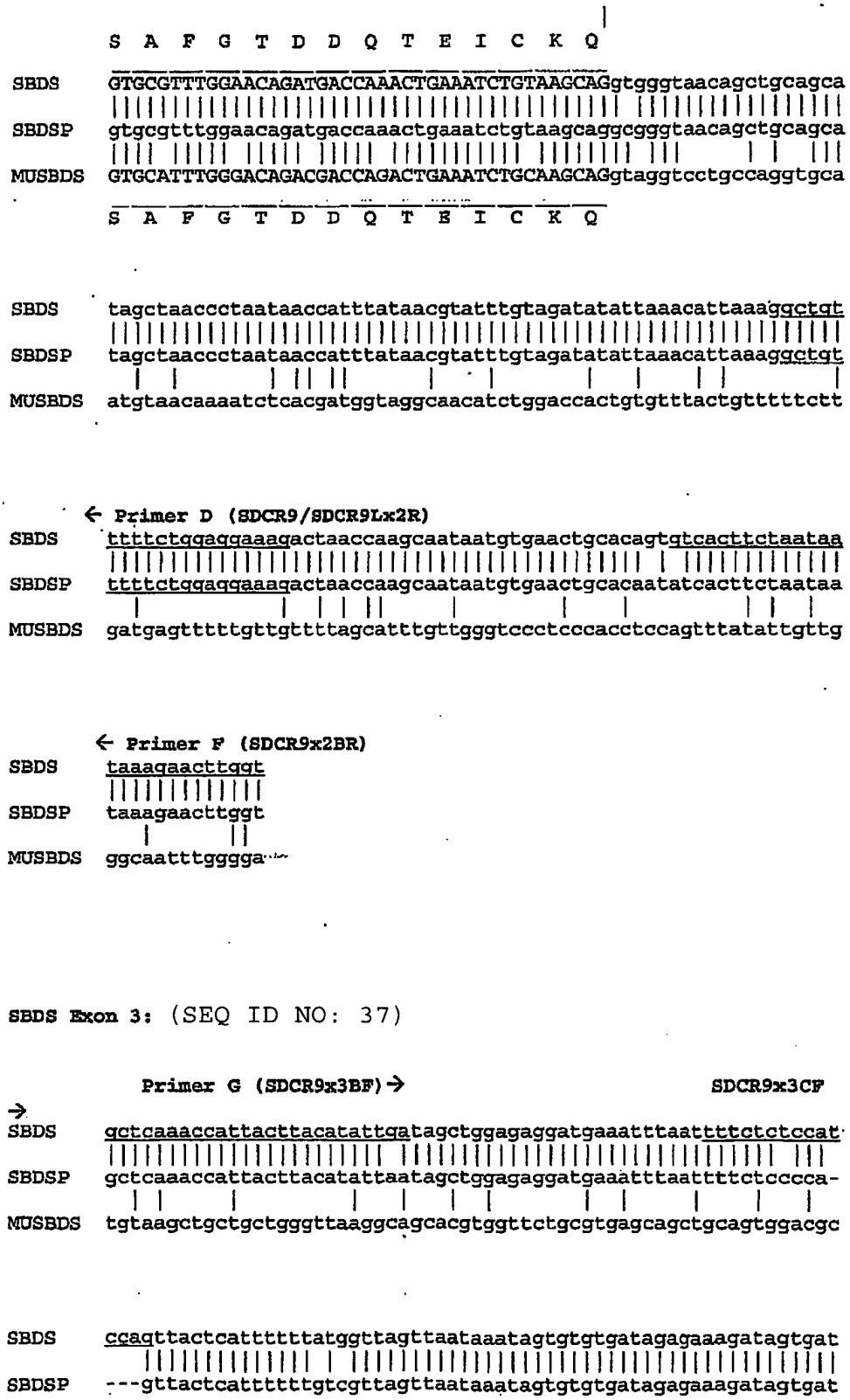
Fig. 6 (cont.)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S A F G T D D Q T E I C K Q
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MUSBDS atgtaacaaaatctcacgatggtaggcaacatctggaccactgtgtttactgtttttctt
← Primer D (SDCR9/SDCR9Lx2R)
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← Primer F (SDCR9x2BR)
SBDS taaaqaacttqqt
SBDSP taaagaacttggt
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Primer G (SDCR9x3BF)→
SDCR9x3CF
→
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SBDSP gctcaaaccattacttacatattaatagctggagaggatgaaatttaattttctcccca-
MUSBDS tgtaagctgctgctgggttaaggcagcacgtggttctgcgtgagcagctgcagtggacgc
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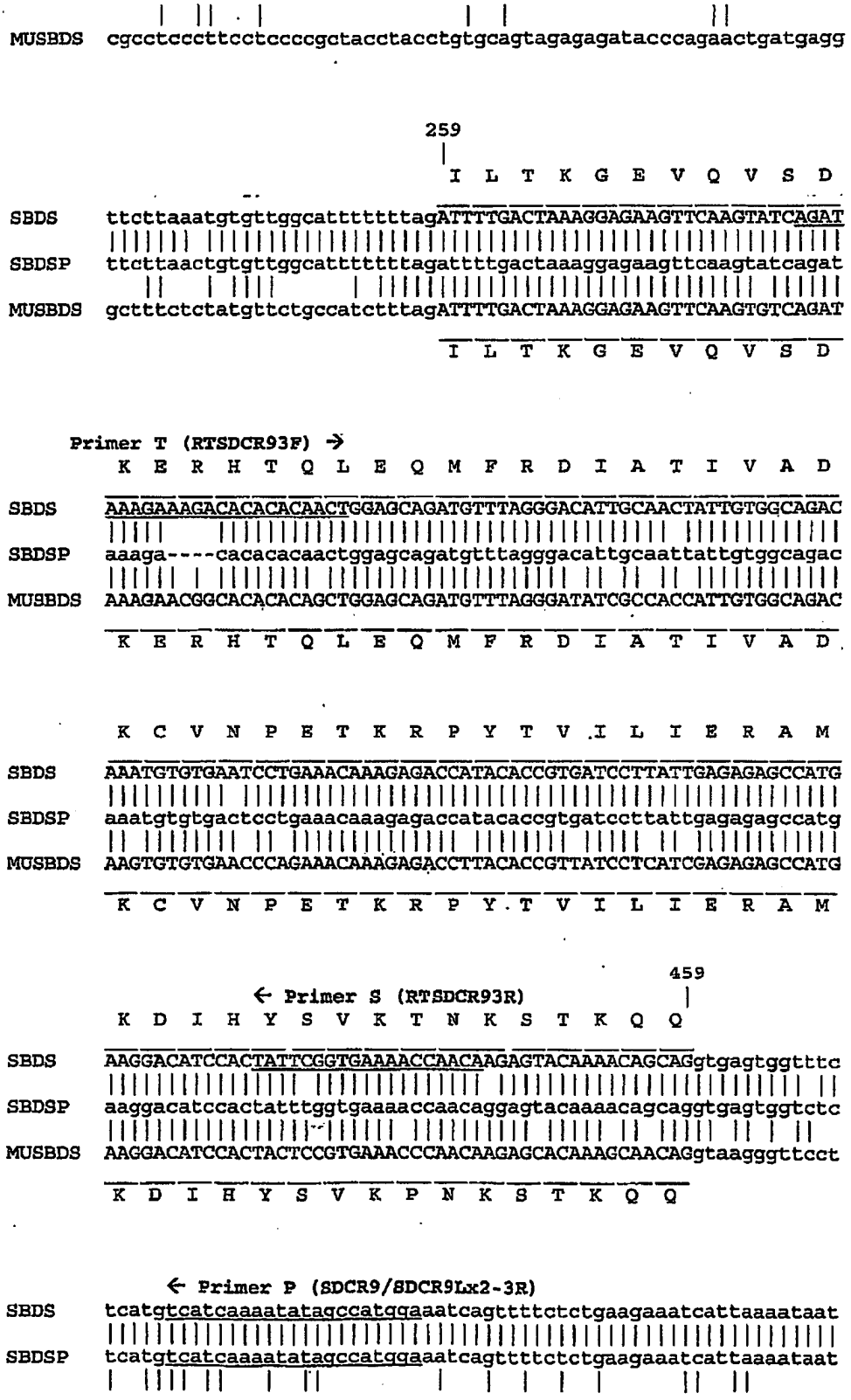
Fig. 6 (cont.)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SBDSP aaaga----cacacacaactggagcagatgtttagggacattgcaattattgtggcagac
MUSBDS AAAGAACGGCACACACAGCTGGAGCAGATGTTTAGGGATATCGCCACCATTGTGGCAGAC
K E R H T Q L E Q M F R D I A T I V A D
K C V N P E T K R P Y T V I L I E R A M
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MUSBDS AAGTGTGTGAACCCAGAAACAAAGAGACCTTACACCGTTATCCTCATCGAGAGAGCCATG
K C V N P E T K R P Y T V I L I E R A M
459
← Primer S (RTSDCR93R)
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SBDS AAGGACATCCACTATTCGGTGAAAACCAACAAGAGTACAAAACAGCAGgtgagtggtttc
SBDSP aaggacatccactatttggtgaaaaccaacaggagtacaaaacagcaggtgagtggtctc
MUSBDS AAGGACATCCACTACTCCGTGAAACCCAACAAGAGCACAAAGCAACAGgtaagggttcct
K D I H Y S V K P N K S T K Q Q
← Primer P (SDCR9/SDCR9Lx2-3R)
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Fig. 6 (cont.)

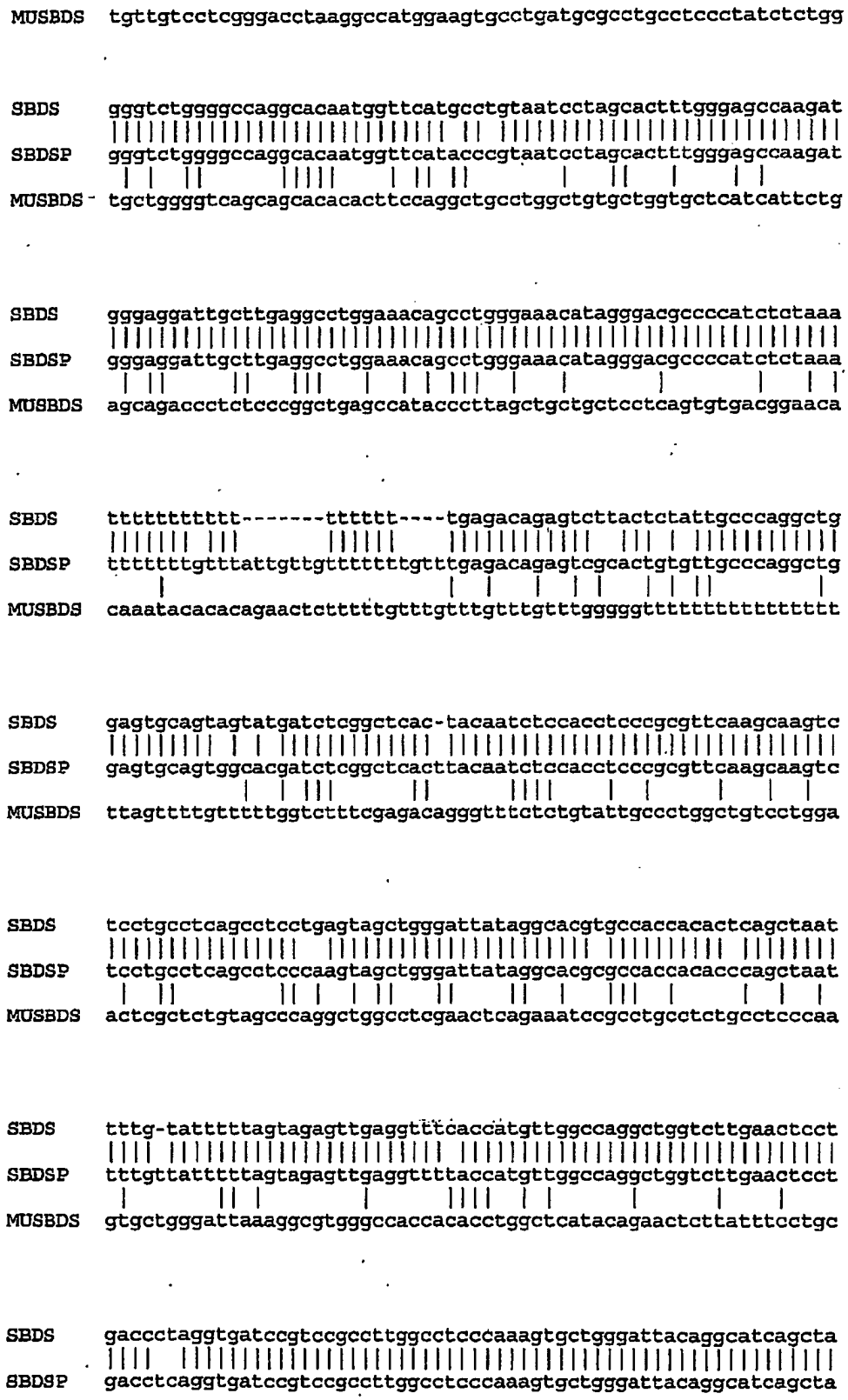
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Fig. 6 (cont.)

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MUSBDS  tgtgtttattaacatatttcctacagctcagccctgtcacgccagccattctgctggcct

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SBDSP   aaagggtcatttttaacacctcttttttgaatttttcaatttacatataattcacatacaat
MUSBDS  ctcaaaagaaataacaagtcgggtgtggtggtgcacacctttaatcccagcactcgggag

SBDS    aaatttcacactcataaagtatgtacactttaagtggtatattaacaaagttttggaacc
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SBDS    ttccctgctacctggttcgagaacattttcatcaccacaaaaagaaagtcagtatccatt
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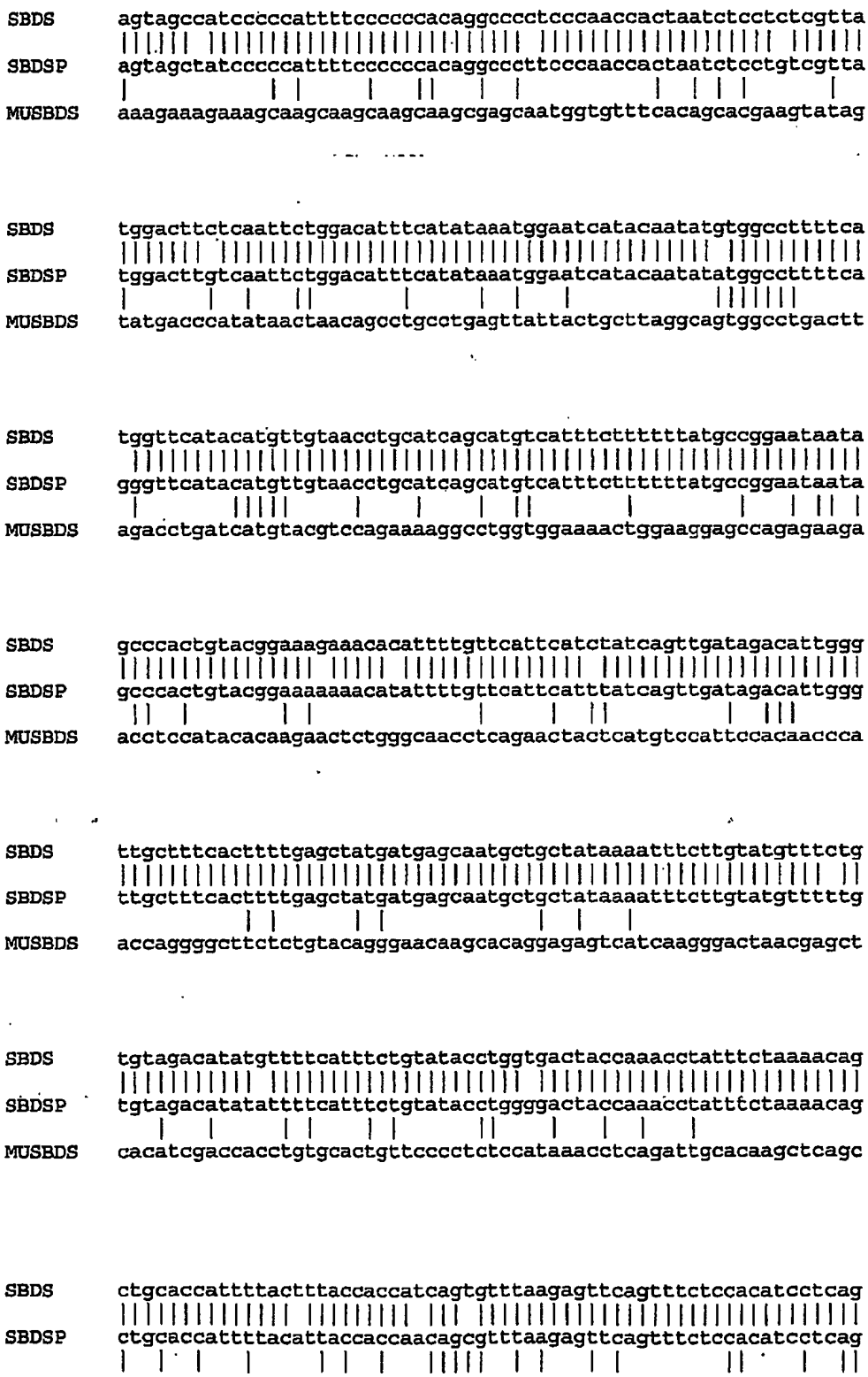
Fig. 6 (cont.)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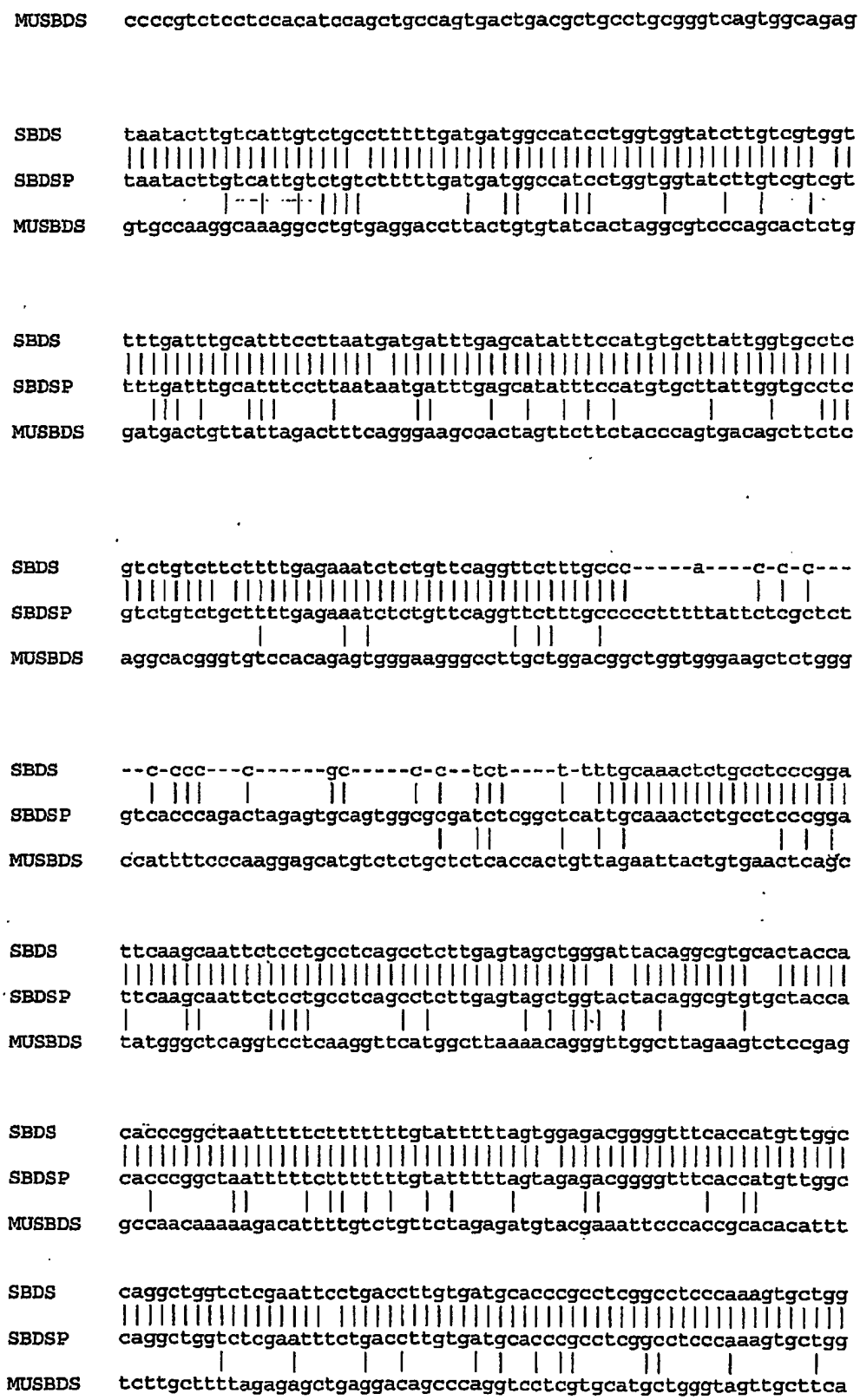
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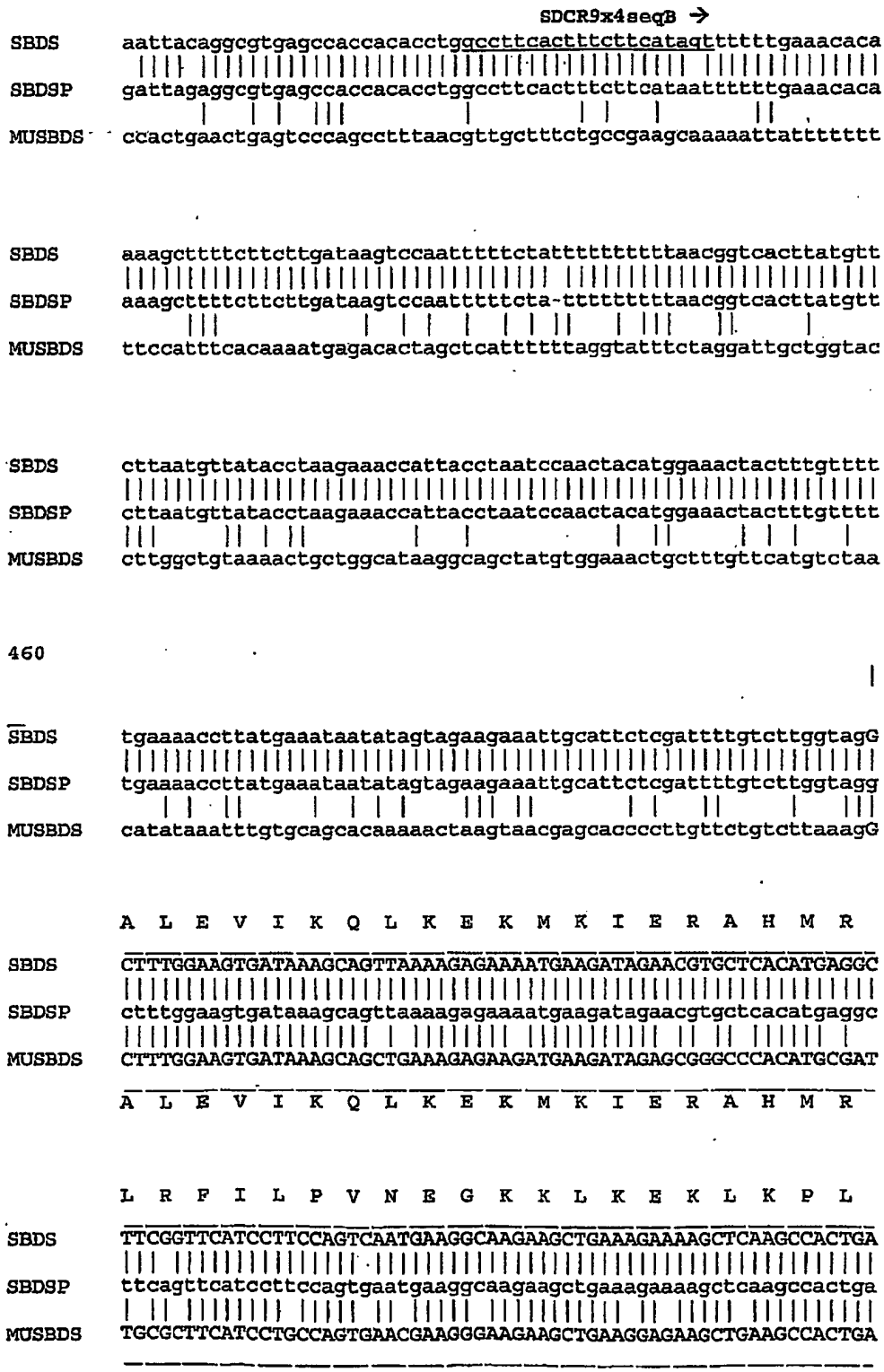
Fig. 6 (cont.)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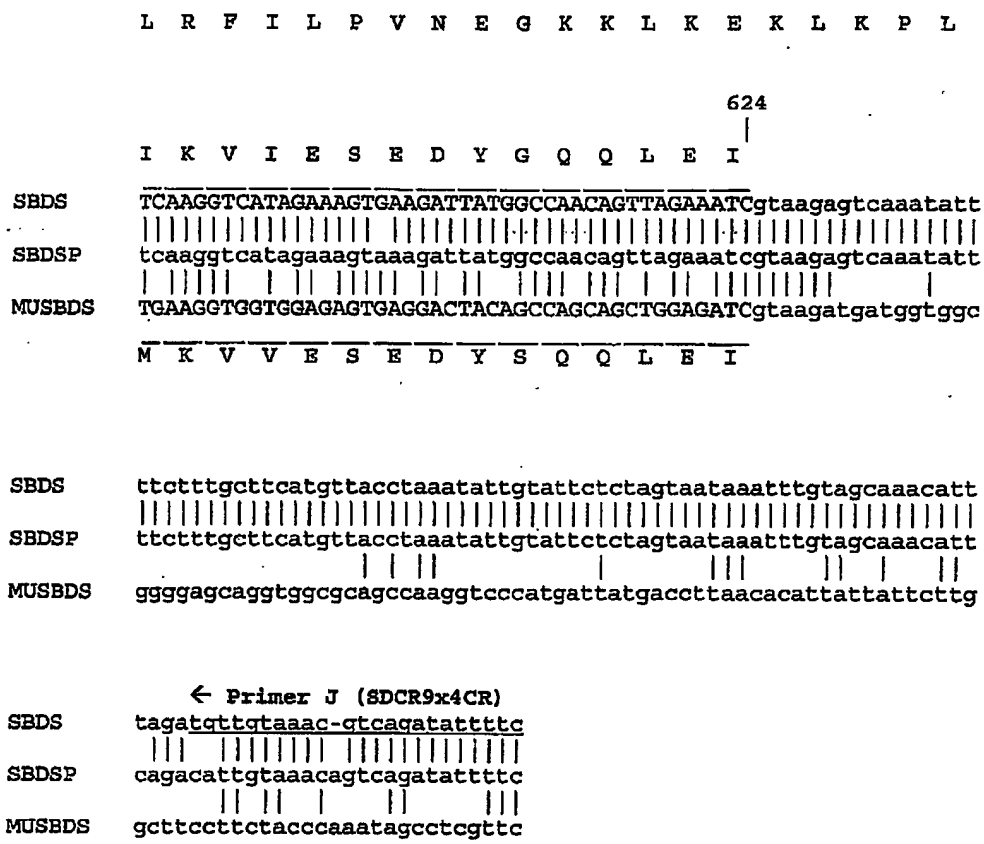

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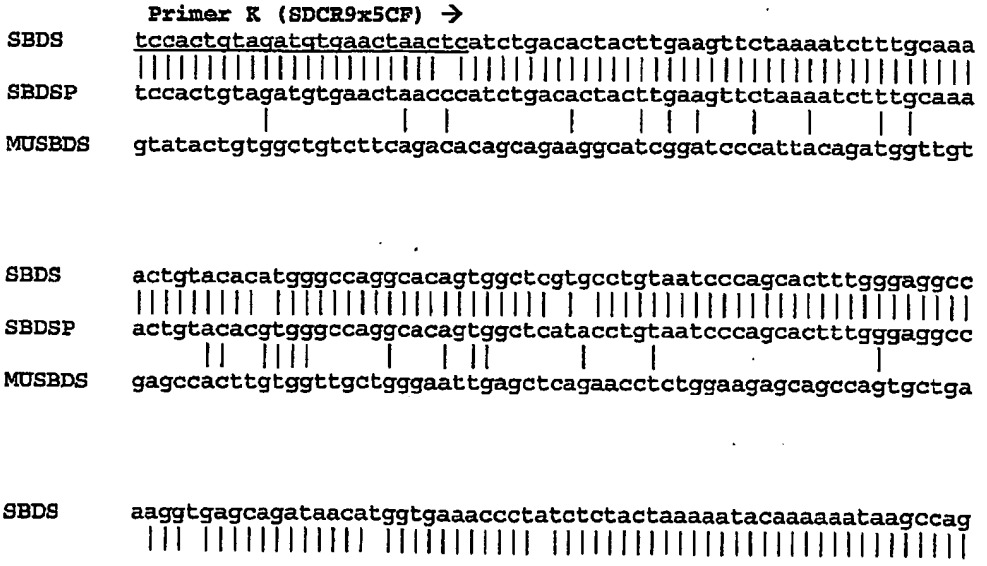

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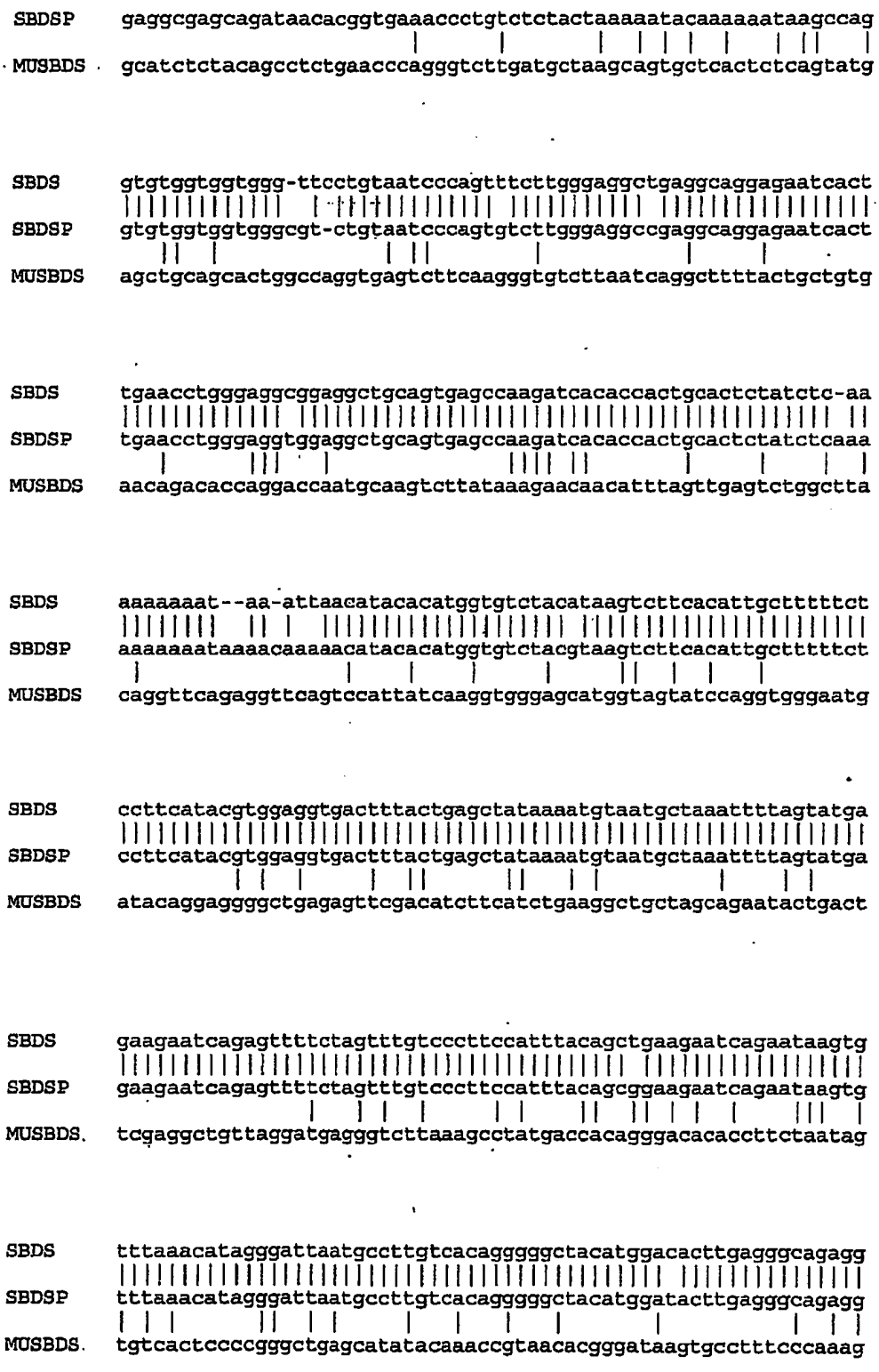
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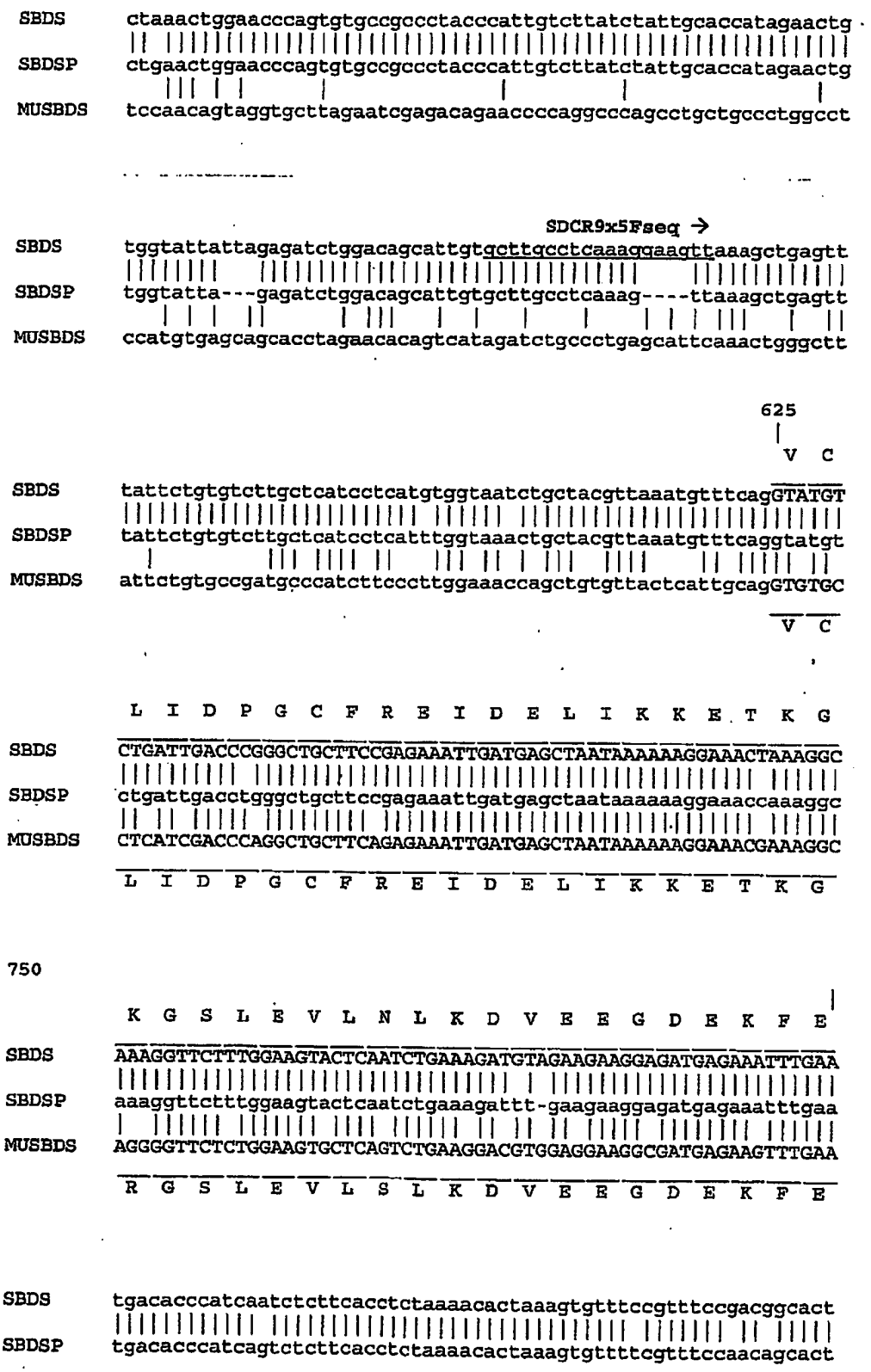
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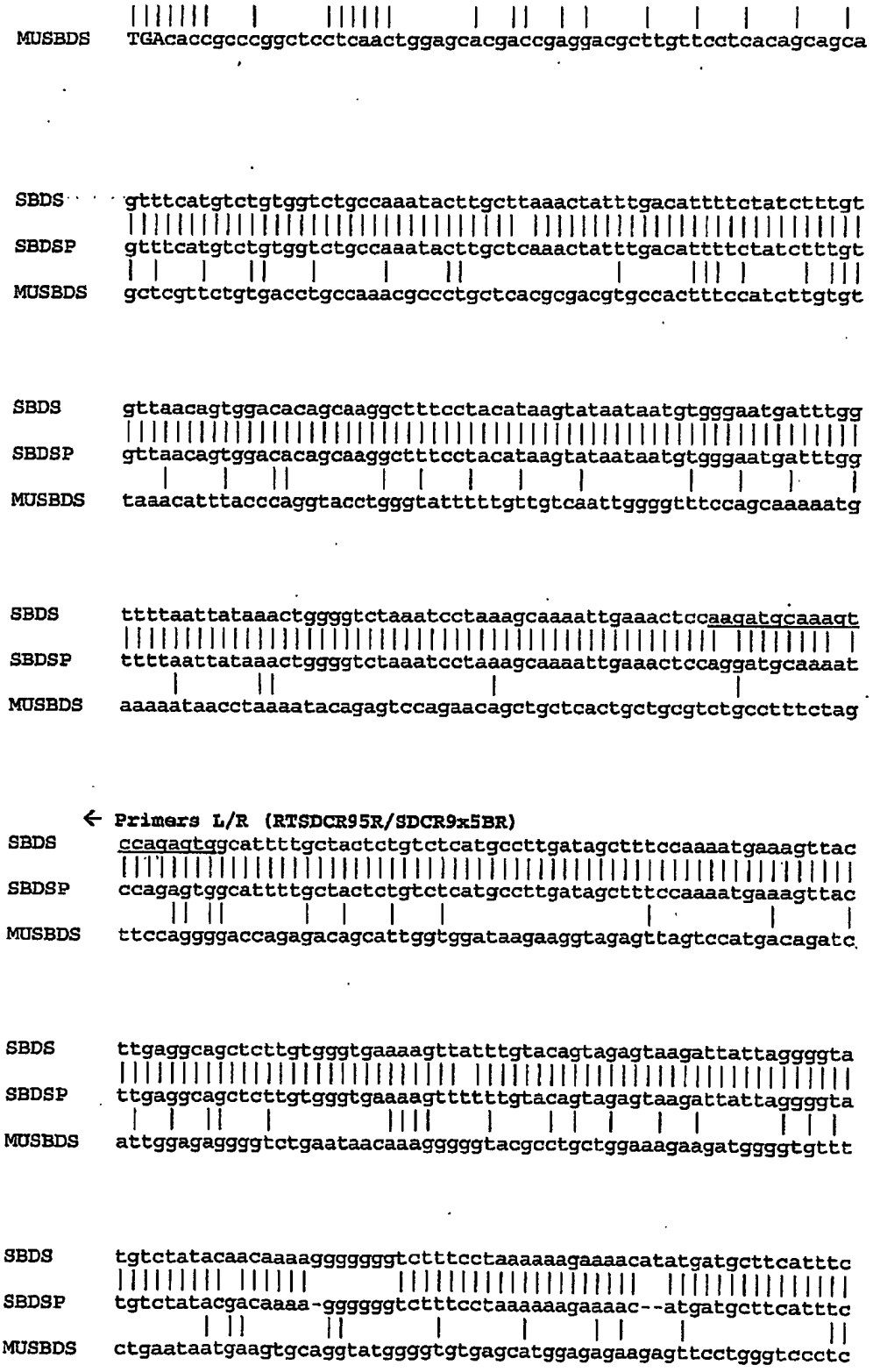
Fig. 6 (cont.)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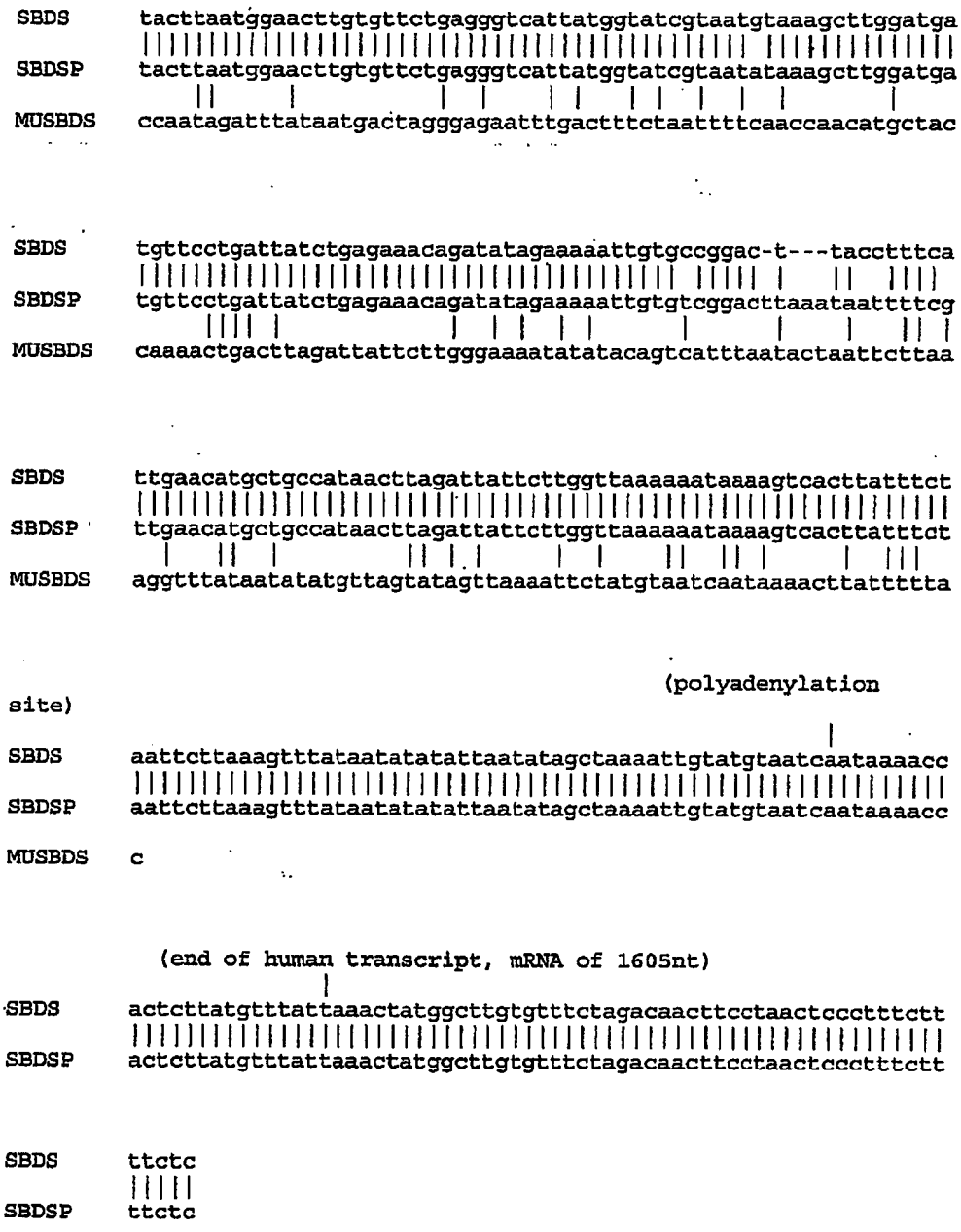
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Fig. 6 (cont.)